

## The Future of Genetic Testing

### Telling Science Fact from Science Fiction

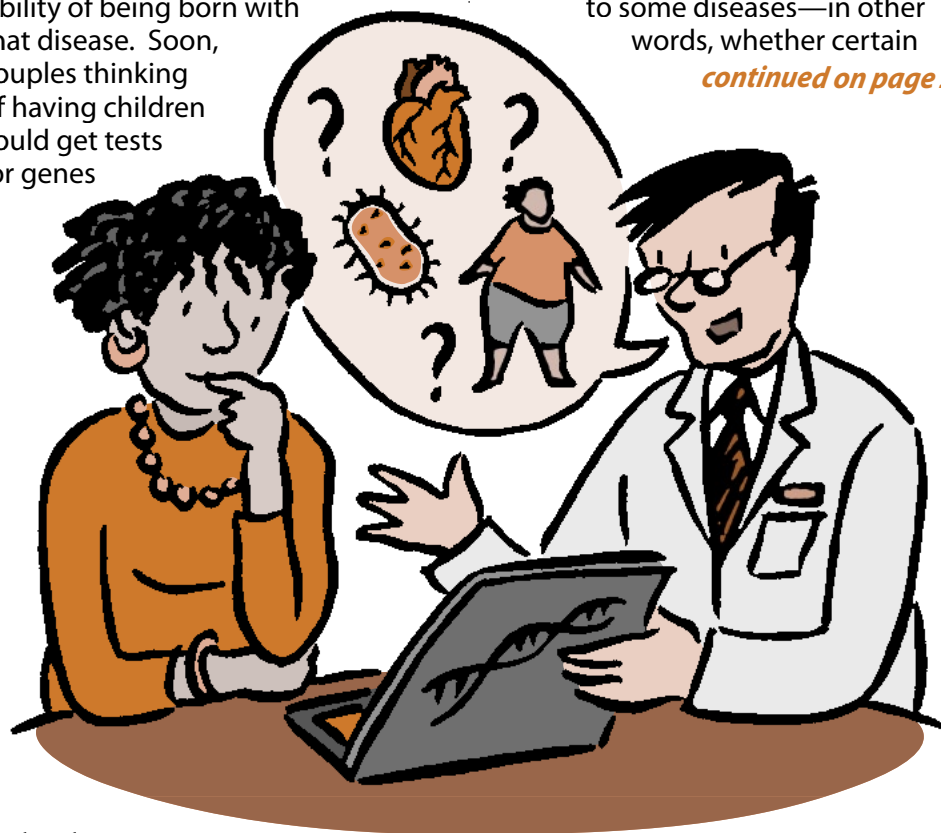
Are you likely to get heart disease? Is obesity in your future? Your risk of developing many diseases and health conditions is partly written in your **genes**. One day soon you'll be able to visit the doctor, have some blood drawn and find out more about your health risks for the next 5 or 10 years through a method called **genetic testing**. But we still have many things to learn about genes before that vision becomes a reality.

First, the facts: The idea of genetic testing isn't new. As early as the 1960s, doctors began urging that newborn babies be tested for rare diseases they could inherit from their parents. Such diseases are called genetic diseases because the illnesses are contained in genes. Testing began for phenylketonuria (PKU), a rare disease that causes mental retardation. PKU can be prevented with a special diet if it's detected early. The test for PKU and other rare but treatable diseases are now routinely done in the hospital soon after a baby is born.

In the 1970s, researchers developed genetic tests that could be done before a baby is born. Scientists found that if a mother and father both have the gene for a certain illness, their child has a high possibility of being born with that disease. Soon, couples thinking of having children could get tests for genes

People with certain forms of a gene called BRCA-1, for example, are more likely to develop breast cancer. Genetic testing of this type can help you find out whether you have a "genetic predisposition" to some diseases—in other words, whether certain

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### Definitions

#### Genes

Stretches of DNA, a substance you inherit from your parents, that define characteristics like height and eye color, along with how likely you are to get certain diseases.

#### Genetic Testing

Test that involves taking a sample from someone's blood, hair, skin or other body part, then examining the person's genes to look for signs they might be at high risk for certain diseases or disorders.

that put their children at risk for developing sickle cell disease and Tay Sach's. By the 1980s, tests called prenatal genetic tests were regularly done before a baby was born to find out the risk of a genetic disease. They're now a routine part of health care in most states.

In the 1990s, researchers were able to identify genes that showed a person's chance of developing breast cancer and colon cancer.

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## Genetic Testing and Counseling:

[www.genome.gov/10002335](http://www.genome.gov/10002335)

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gene variations you carry make you more likely to develop a disease. That doesn't mean that you're destined to develop it; it only means you're more likely than those with other versions of the gene.

"It's important to realize that nearly every disease has both a genetic component and an environmental component," Dr. Benjamin Wilfond of NIH's National Human Genome Research Institute explains. "You can't modify your genes, but you can modify your environment to help prevent some diseases."

Consider the common cold virus, for example. Anyone who comes in

contact with the virus could become infected. However, some people have a genetic predisposition that makes them more likely to get sick. If you know this ahead of time, you can take measures like steering clear of places where cold germs are likely to be and taking extra care to wash your hands when visiting public places.

Genetic testing can give us a similar head start on fighting many diseases. People who learn early that they're genetically predisposed to a disease can benefit by knowing what symptoms to look for and potentially finding the disease in its early stages. They may also be able to change things in their lifestyle—their environment—to prevent the disease.

By the early 2000s, computers and other technology had advanced to the point where large collections of a person's genes could be looked at, a method called genetic profiling. In



## Definitions

### Genome

All the genes in your body.

the future, some technology companies may be able to offer genetic testing to map your whole **genome**—all the genes in your body at once! That sounds like a dream come true for preventing disease, but the reality is that researchers haven't yet learned enough to help predict your health.

"The marketing for some testing may be premature," Dr. Wilfond warns. "We know so little about some of these genes, and they may be small contributors to actual disease. There is a danger of misunderstanding or misinterpreting the results. These tests are giving you just small amounts of information."

Wilfond advises people to think carefully about offers for testing. "Ask yourself a couple of questions before you decide," he says. "Is this test part of routine medical care that will help me, or is this test something that's only going to make somebody a lot of money?"

In the meantime, researchers are learning more about genetics and disease every day. "We now understand how to look at a person's genes for their risks of more common diseases such as heart disease, asthma and Parkinson's disease," Wilfond points out. As scientists learn more about how environmental factors affect genes and how illnesses develop, more types of genetic testing will become part of routine health care.

Television and movies—and even some marketing—often make the science fiction of genetic testing seem like science fact. It's true that health researchers are always looking far ahead to a day in science future. Part of the challenge of genetic testing is figuring out what to tell people until that day comes.

"There is a process for accepting new things in medicine that happens to be gradual and that's good," Wilfond observes, "because as we wait longer, we learn more." ■



## Wise Choices

### Should You Get a Genetic Test?

Considering whether to have a genetic test is a lot like deciding whether to have any other medical test, according to Dr. Benjamin Wilfond of NIH's National Human Genome Research Institute. You should ask yourself—and then your doctor—a couple of questions first:

- **Is this test a routine part of medical care?** A test that has

already become part of regular health care practice is well known to doctors and other people on your medical team.

#### ■ What will the results tell me?

As with any test, it's important to have the results reviewed by experts who understand them. Doctors will often recommend you consult a genetic counselor, a type of health professional with expertise in medical genetics and counseling, to help you make decisions about your health based on the test results.

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# Genes or Environment?

## Epigenetics Sheds Light on Debate

Which is more important in shaping who we are and what we will become—our **genes** or the environment around us? For centuries, people have debated whether nature or nurture decides how we look and act. Now, a field of research called **epigenetics** is showing that we can't really separate one from the other.

"We can no longer argue whether genetics or environment has a greater impact on our health and development," argues Dr. Randy Jirtle, a genetics researcher in Duke University's Department of Radiation Oncology, "because both are inextricably linked." Jirtle recently co-chaired a conference on epigenetics that was co-sponsored by Duke and NIH's National Institute of Environmental Health Sciences (NIEHS).

Your body "reads" genes and follows their instructions to make you who you are. Epigenetics is the study of factors that can change the way those genes are read without changing the genetic code itself. According to Jirtle, nutrients, toxins or other things in the environment can cause such "epigenetic" changes. They are one reason, for example, why identical twins who share the same set of genes can be so different.

Epigenetics can have long-lasting effects. Even poor nutrition during pregnancy can cause epigenetic changes that affect a child's risk of getting many diseases later in life.



### Definitions

#### Epigenetics

The study of factors that affect genes in a lasting way without modifying the genetic code itself.

#### Genes

Stretches of DNA, a substance you inherit from your parents, that define characteristics like height and eye color.

Dr. David Barker of Oregon Health and Sciences University in Portland, Oregon says, "Studies around the world have shown that people with low birth weight have significantly increased risk of coronary heart disease, stroke, type 2 diabetes, hypertension and osteoporosis."

Research shows that these epigenetic changes can even be passed down from one generation to the next.

Scientists are now discovering exactly how environmental factors produce these lasting changes. A major way genes can be altered is by something called methylation. That's when a bunch of atoms scientists call a methyl group gets chemically attached to a gene.

Dr. Frederick Tyson, a program administrator with NIEHS and co-organizer of the conference, explains, "The addition of the methyl group 'silences' the gene...." In other words, when a gene's had a methyl group attached to it, your body can't read it anymore, even though the genetic code hasn't changed one bit.

Studies by Jirtle's research team have shown that epigenetic changes like this can change the course of development. In one set of experiments, pregnant mice eating a diet with four common nutritional supplements gave birth to mice with brown or black fur. Pregnant mice eating the same diet without the supplements gave birth to mice with yellow coats.

Even more important, the mice whose mothers received the supplements had less obesity, diabetes and cancer as adults. The researchers went on to discover that the nutrients affected a gene called *agouti*,



leading to both the changes in coat color and susceptibility to disease.

NIEHS's research programs now aim to learn more about how environmental factors influence genes and affect your risk for disease. Investigators are looking at a wide range of substances in the environment, including pesticides and heavy metals, to see which can alter genes and which genes they're altering.

At least a dozen drugs targeting epigenetic changes are already being tested, and more are in development. Last May, the Food and Drug Administration approved a drug that affects methylation to treat a rare bone marrow disorder that can lead to leukemia. As researchers learn more about how genes and environmental factors interact, they'll be able to develop new approaches for disease prevention and treatment. ■



## Health Capsules

### SIDS Risk Up in Winter

Do you put extra blankets or clothes on your infant during the cold winter months, hoping to keep him or her warmer? The extra layers may actually increase the risk for Sudden Infant Death Syndrome (SIDS), the sudden, unexplained death of an infant in the first year of life. The number of infants who die from SIDS goes up during the winter, according to NIH's National Institute of Child Health and Human Development (NICHD).

For more than a decade, NICHD has led the "Back to Sleep" campaign, which explains how to reduce the risk of SIDS. Since the campaign began, the overall SIDS rate in the U.S. has gone down by more than 50%. Despite the campaign's progress, however, SIDS is the leading cause of death in infants between 1 month and 1 year of age, claiming the lives

of about 2,500 each year.

Most SIDS deaths happen between 2 and 4 months of age. While the causes of SIDS are still unclear, you can reduce factors that increase SIDS risk. NICHD endorses these recommendations from the American Academy of Pediatrics:

- Always place your baby on his or her back to sleep.
- Place your baby on a firm sleep surface, such as a safety-approved crib mattress, covered by a fitted sheet.
- Keep soft objects, toys and loose bedding out of the sleep area.
- Don't allow smoking around your baby.
- Keep your baby's sleep area close to, but separate from, where you and others sleep.
- Consider offering a clean, dry pacifier when placing your baby on his or her back to sleep.

- Don't let your baby overheat during sleep. The temperature should be kept at a level that feels comfortable for an adult.
- Avoid products that claim to reduce the risk of SIDS.
- Don't use home monitors to reduce the risk of SIDS.
- Reduce the chance that flat spots will develop on your baby's head by providing "Tummy Time" when your baby is awake and someone is watching; changing the direction that your baby lies in the crib; and avoiding too much time in car seats, carriers and bouncers.

NICHD has a variety of free "Back to Sleep" education materials available for parents, caregivers and health care providers. Many are available in English and Spanish. ■



[www.nichd.nih.gov/sids](http://www.nichd.nih.gov/sids)  
or 1-800-370-2943

### Risky Teen Behavior

Three of the leading causes of preventable death in the U.S. are smoking, being overweight or obese, and abusing alcohol. A new study shows that a large proportion of American youth are already involved in these risky health behaviors.

NIH-funded researchers at the Carolina Population Center and the University of North Carolina at Chapel Hill carried out the study. They used data from the National Longitudinal Study of Adolescent Health, which was designed to measure the effects of home, family and school environments on behaviors that promote health. It's the most comprehensive survey of adolescent health behavior to date.

More than 14,000 high school and middle school students from around the country were surveyed for the study. Researchers first interviewed them when they ranged from 12 to 19 years of age and then again when they were 19 to 26 years old. The students answered questions about

diet, exercise, tobacco use, substance use, binge drinking, violence, reproductive health, mental health and access to health care.

Overall, the study found that many health behaviors got worse during the transition to young adulthood. There were "dramatic" increases in some of the biggest contributors to preventable deaths: smoking, poor diet and physical inactivity, and alcohol consumption.

On the plus side, there was less depression at young adulthood than at the younger ages. Young adults were also less likely to have suicidal thoughts or be involved in violence.

The researchers found significant differences between racial groups. These differences became even greater in young adulthood. The finding that racial groups differ in their health behaviors means that one-size-fits-all intervention programs may not be effective. These findings can now help guide the creation of more effective intervention programs tailored to particular groups. ■



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